

Table 5. ASPM Mutations

cDNA	Mutation class	Mutation Subtype	Predicted Effect	Occurrence
Translocation	Translocation			1
Large deletion	Deletion		no ASPM production	1
c.74delG	Deletion	Frame shift	p.Arg25fs	1
c.77delG	Deletion	Frame shift	p.Gly26fs	1
c.297+1460_3391-242del21844	Deletion		Loss of microtubular binding domain	1
c.349C>T	Substitution	Stop mutation	p.Arg117X	4
c.440delA	Deletion	Frame shift	p.Lys147fs	1
c.577C>T	Substitution	Stop mutation	p.Gln193X	1
c.719_720delCT	Deletion	Frame shift	p.Ser240fs	1
C1002delA	Deletion	Frame shift	p.Val335fs	1
c.1152_1153delAG	Deletion	Frame shift	p.Ser384fs	1
c.1179delT	Deletion	Frame shift	p.Pro393fs	1
c.1258_1264delTCTCAAG	Deletion	Frame shift	P.Ser420fs	4
c.1260_1266delTCAA GTC	Deletion	Frame shift	p.Gln421fs	1
c.1366G>T	Substitution	Stop mutation	p.Glu456X	1
c.1406_1413delATCC TAAA	Deletion	Frame shift	p.Asn469fs	1
c.1590delA	Deletion	Frame shift	p.Lys530fs	1
c.1628_1632delCTTAT	Deletion	Frame shift	p.Ser543fs	1
c.1630_1634delATCTT	Deletion	Frame shift	p.Tyr544fs	1
c.1727_1728delAG	Deletion	Frame shift	p.Lys576fs	1
c.1959_1961delCAAA	Deletion	Frame shift	p.Asn653fs	1
c.1990C>T	Substitution	Stop mutation	p.Gln664X	1
c.2389C>T	Substitution	Stop mutation	p.Arg797X	1
c.2389C>T and c.7781-7782delAG		Stop mutation	[p.Arg797X] and [p.Gln2594fsX6]	1
c.2761-25A>G	Substitution	Splicing/ Frame shift	create a AG motif	1
c.2936+5G>T	Substitution	Splicing/ Frame shift	affects splice donor site	1
c.2938C>T	Substitution	Stop mutation	p.Arg980X	1
c.2967G>A	Substitution	Stop mutation	p.Trp989X	1
c.3055C>T	Substitution	Stop mutation	p.Arg1019X	1
c.3082G>A	Substitution	Splicing/ Frame shift	affects splice donor site	1
c.3188T>G	Substitution	Stop mutation	p.Leu1063X	1
c.3477_3481delCGCT A	Substitution	Stop mutation	p.Ala1160fs	1
c.3527C>G	Substitution	Stop mutation	p.Thr1176X	1
c.3663delG	Deletion	Frame shift	p.Arg1221fs	2

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c.3710C>G	Substitution	Stop mutation	p.Ser1237X	1
c.3741+1G>A	Substitution	Splicing/ Frame shift	affects splice donor site	1
c.3796G>T	Substitution	Stop mutation	p.Glu1266X	1
c.3811C>T	Substitution	Stop mutation	p.Arg1271X	4
c.3978G>A	Substitution	Stop mutation	p.Trp1326X	2
c.4074G>A	Substitution	Stop mutation	p.Trp1358X	1
c.4195insA	Insertion/duplication	Frame shift	p.Thr1399fs	1
c.4581delA	Deletion	Frame shift	p.Gly1527fs	1
c.4795C>T	Substitution	Stop mutation	p.Arg1599X	1
c.4855_4856delTA	Deletion	Frame shift	p.Tyr1619fs	1
c.5136C>A	Substitution	Stop mutation	p.Tyr1712X	1
c.5149delA	Deletion	Frame shift	p.Ile1717fs	1
c.6189T>G	Substitution	Stop mutation	p.Tyr2063X	1
c.6232T>C	Substitution	Stop mutation	p.Arg2078X	1
c.6335_6336delAT	Deletion	Frame shift	p.His2112fs	1
c.6651_6654delAACAA	Deletion	Frame shift	p.thr2218fs	1
c.6732delA	Deletion	Frame shift	p.Tyr2245fs	1
c.7489_7493delTATAT	Deletion	Frame shift	p.Tyr2497fs	1
c.7761T>G	Substitution	Stop mutation	p.Tyr2587X	2
c.7782_7783delGA	Deletion	Frame shift	p.Lys2595fs	2
c.7859_7860delAG	Deletion	Frame shift	p.Gln2620fs	1
c.7894C>T	Substitution	Stop mutation	p.Gln2632X	1
c.8130_8131delAA	Deletion	Frame shift	p.Thr2710fs	1
c.8190_8191delAG	Deletion	Frame shift	p.Glu2731fs	1
c.8273T>A	Substitution	Stop mutation	p.Leu2758X	1
c.8378delT	Deletion	Frame shift	p.Met2793fs	1
c.8508_8509delGA	Deletion	Frame shift	p.Gln2836fs	3
c.8668C>T	Substitution	Stop mutation	p.Gln2890X	1
c.8844delC	Deletion	Frame shift	p.Ala2948fs	1
c.9118_9119insCATT	Insertion/Duplication	Frame shift	p.Tyr3040fs	1
c.9159delA	Deletion	Frame shift	p.Lys3053fs	2
c.9178C>T	Substitution	Stop mutation	p.Gln3060X	3
c.9190C>T	Substitution	Stop mutation	p.Arg3064X	2
c.9238A>T	Substitution	Stop mutation	p.Leu3080X	2
c.9319C>T	Substitution	Stop mutation	p.Arg3107X	1
c.9492T>G	Substitution	Stop mutation	p.Tyr3164X	1
c.9507delG	Deletion	Frame shift	p.Ile3170fs	1
c.9539A>C	Substitution	Missense mutation	p.Gln3180Pro	1
c.9557C>G	Substitution	Stop mutation	p.Ser3186X	3
c.9595A>T	Substitution	Stop mutation	p.Lys3199X	1

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c.9677_9678insG	Deletion	Frame shift	p.Cys3226fs	1
c.9681delA	Deletion	Frame shift	p.Thr3227fs	1
c.9686_9690delTTAAA	Deletion	Frame shift	p.Ile3229fs	1
c.9697C>T	Substitution	Stop mutation	p.Arg3233X	1
c.9730C>T	Substitution	Stop mutation	p.Arg3244X	1
c.9745_9746delCT	Deletion	Frame shift	p.Leu3249fs	1
c.9754delA	Deletion	Frame shift	p.Arg3252fs	1
c.9789T>A	Substitution	Stop mutation	p.Tyr3263X	1
c.9841A>T	Substitution	Stop mutation	p.Arg3281X	1
c.9984+1G>T	Substitution	Splicing/Frame shift	affects splice donor site	1
c.10059G>A	Substitution	Stop mutation	p.Tyr3353X	1

Mutations previously reported and personal data